

Radiological imaging of Hirschsprung disease.



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Hirschsprung disease is the most common cause of neonatal colonic obstruction (15-20%). It is commonly characterized by a short segment of colonic aganglionosis affecting term neonates, especially boys.

Epidemiology

Hirschsprung disease affects approximately 1:5000-8000 live births. In short segment disease, there is a significant predilection for males (M:F of ~3.5:1), which reduces with increasing length of involvement. Interestingly, it is almost never seen in premature infants.

Clinical presentation

The condition typically presents in term neonates with failure to pass meconium in the first 1-2 days after birth, although later presentation is also common. Overall ~75% of cases present within six weeks of birth, and over 90% of cases present within the first five years of life. A very small number may present in the adult population.

In cases of delayed presentation with anorectal constipation, manometry may be useful in distinguishing short/ultra short segment Hirschsprung disease from other causes.

A definitive diagnosis requires a full thickness rectal biopsy.

Pathology

Hirschsprung disease is characterized by aganglionosis (absence of ganglion cells) in the distal colon and rectum. It is thought to either occur from a failure of neuroblasts in neural crest cells to migrate into bowel segments or degeneration of already migrated neuroblasts. It affects cells both in the myenteric and submucosal plexuses. Hence, functional obstruction develops as a result of a spasm in the denervated colon. It can be anatomically divided into four types according to the length of the aganglionic segment:

*short segment disease: ~75% **

rectal and distal sigmoid colonic involvement only

long segment: ~15%

typically extends to splenic flexure / transverse colon

total colonic aganglionosis: ~7.5% (range 2-13%)

also known as Zuezler-Wilson syndrome

occasional extension of aganglionosis into the small bowel

ultrashort segment disease

3-4 cm of internal anal sphincter only

controversial entity

see notes on percentages

It is postulated that hypoganglionosis (reduced number of ganglion cells) handles intestinal pseudo-obstruction.

Associations

Although Hirschsprung is an isolated abnormality in 70% of cases, there are some well-documented associations, including:

Down syndrome: in ~10% of Hirschsprung cases

neurocristopathy syndromes

Waardenburg-Shah syndrome

Haddad syndrome

MEN IIa

neuroblastoma

other non-neurocristopathy syndromes

Aarskog syndrome

Bardet-Biedl syndrome

Fryns syndrome

Pallister-Hall syndrome

Smith-Lemli-Opitz syndrome

Hirschsprung Disease Symptoms:

Eighty percent of children with Hirschsprung disease show symptoms in the first 6 weeks of life. Infants suffering from the disease usually become symptomatic during the first 24 to 48 hours of life. However, children with only a short segment of intestine that lacks normal nerve cells may not show symptoms for several months or even years. While individuals experience a range of symptoms, the following are the most common:

Not having a bowel movement in the first 48 hours of life

Gradual marked swelling of the abdomen

Gradual onset of vomiting

Fever

Children who do not have early symptoms may present with the following:

Sepsis (overwhelming infection)

Constipation that worsens over time

Small, watery stool

Loss of appetite

Delayed growth

Hirschsprung disease diagnosis:

Careful physical examination is required, and physical findings are dependent upon the age at presentation and the severity of the condition. Establishing the diagnosis also includes undergoing a number of diagnostic studies. These include the following:

Abdominal X-ray. This may indicate a bowel blockage. This study only allows physicians to suspect the diagnosis, not definitively diagnose it.

Contrast enema. *This is a procedure performed to examine the large intestine (colon) for abnormalities. A contrast agent is given into the rectum in order to coat the inside of organs so that they will show up on an X-ray. This is the most valuable radiologic study for establishing the diagnosis.*

An X-ray of the abdomen will show a narrowed colon, obstruction and dilated (exceptionally enlarged) intestine above the obstruction.

Rectal biopsy. This procedure will establish the diagnosis of Hirschsprung disease.

A sample of the cells in the rectum is taken and then looked at under a microscope.

Confirmation of Hirschsprung is based on the absence of ganglion cells and the presence of nonmyelinated nerves in the biopsy segment.

In infants, a suction rectal biopsy can be done at the bedside. Since there are no sensory nerves at the site of biopsy, this is not painful. When a suction biopsy is inconclusive, surgical biopsy is performed under general anesthesia in the operating room.

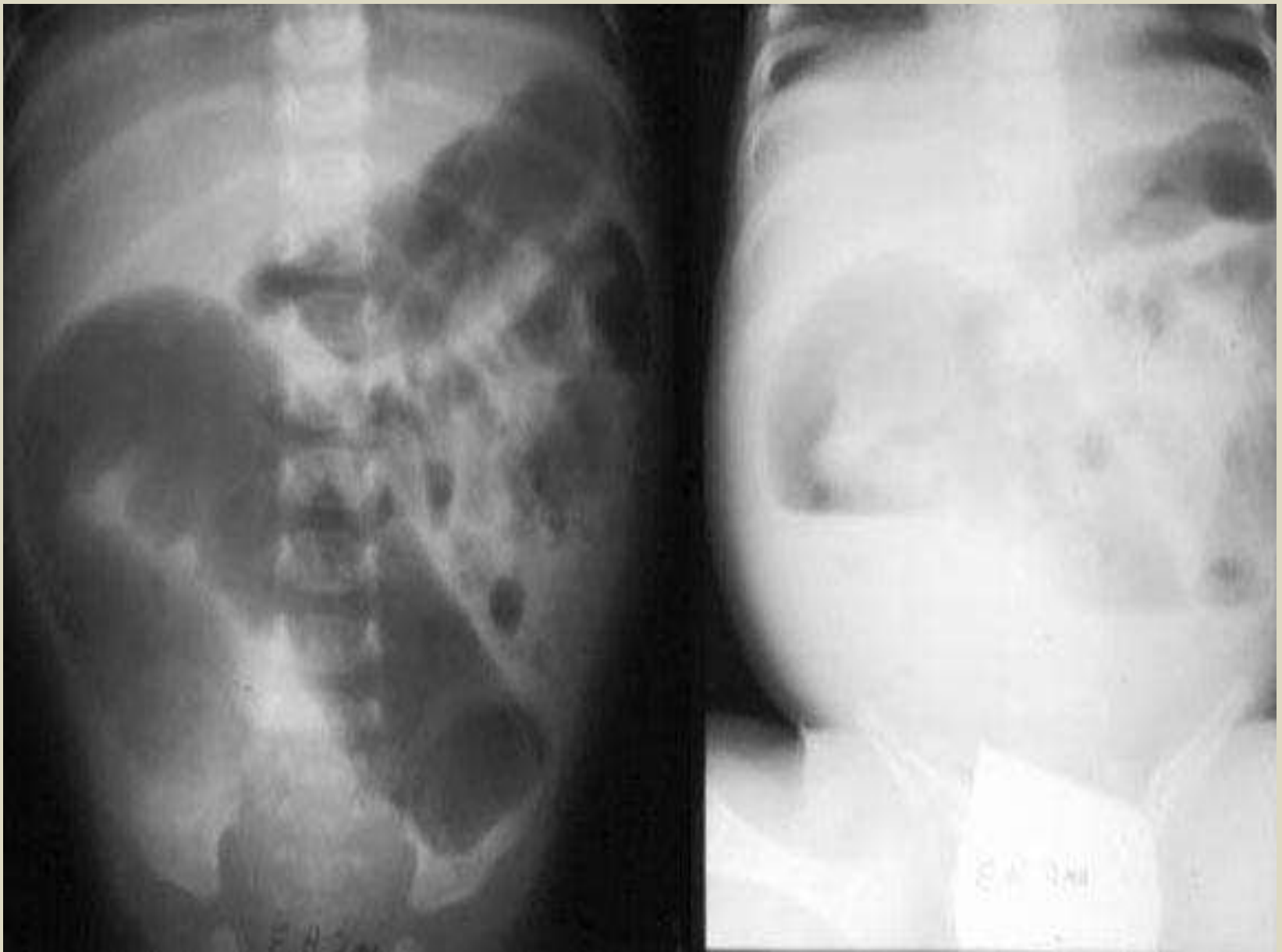
Anorectal manometry. *This determines whether normal reflexes involving the rectum and the anus are present. Used only in older children, the test can be performed at the bedside.*

Radiography

Radiographs of the neonatal abdomen with Hirschsprung disease (HD) may show multiple loops of dilated small bowel with air-fluid levels that can usually be determined to be a distal bowel obstruction. An empty rectum is a common finding. The classic image is a dilated proximal colon with the aganglionic cone narrowing towards the distal gut. A cutoff sign in the rectosigmoid region with an absence of air distally is a useful finding in Hirschsprung-associated enterocolitis (HAEC).



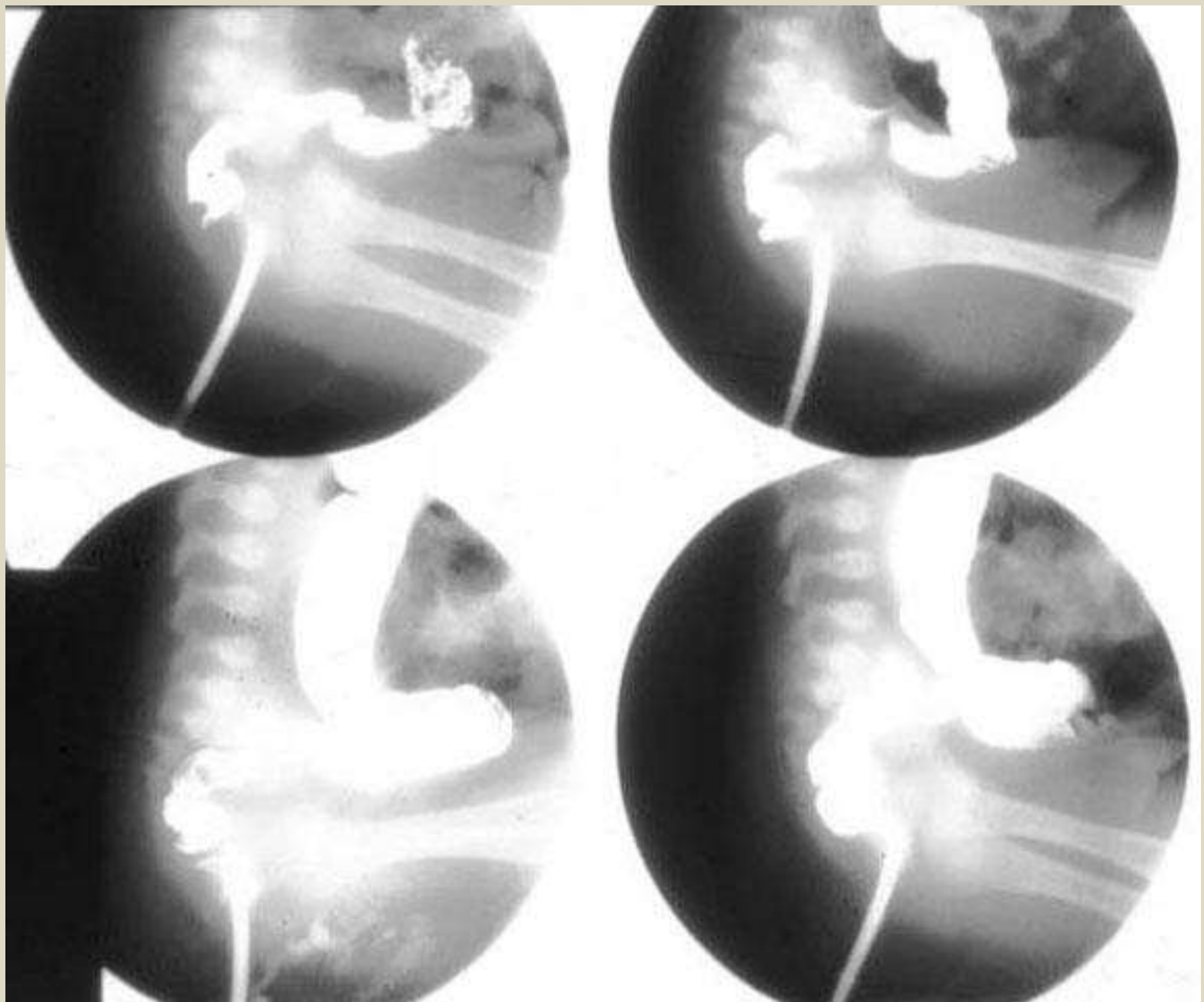
Hirschsprung disease. Frontal abdominal radiograph showing marked dilatation of the small bowel with no gas in the rectum.



Hirschsprung disease. Frontal abdominal radiograph showing marked dilatation of the bowel with no gas in the rectum. In the sitting position, air-fluid levels in the large bowel are seen.



Hirschsprung disease. Lateral abdominal radiograph shows a very enlarged, stool-filled sigmoid. No air or stool content is seen in the rectum.



Hirschsprung disease. Barium enema technique shows slow contrast-material infusion.



Hirschsprung disease. Lateral view from a barium enema examination depicting the reduced diameter of the rectum and sigmoid.



Hirschsprung disease. Barium enema showing reduced caliber of the rectum, followed by a transition zone to an enlarged-caliber sigmoid.



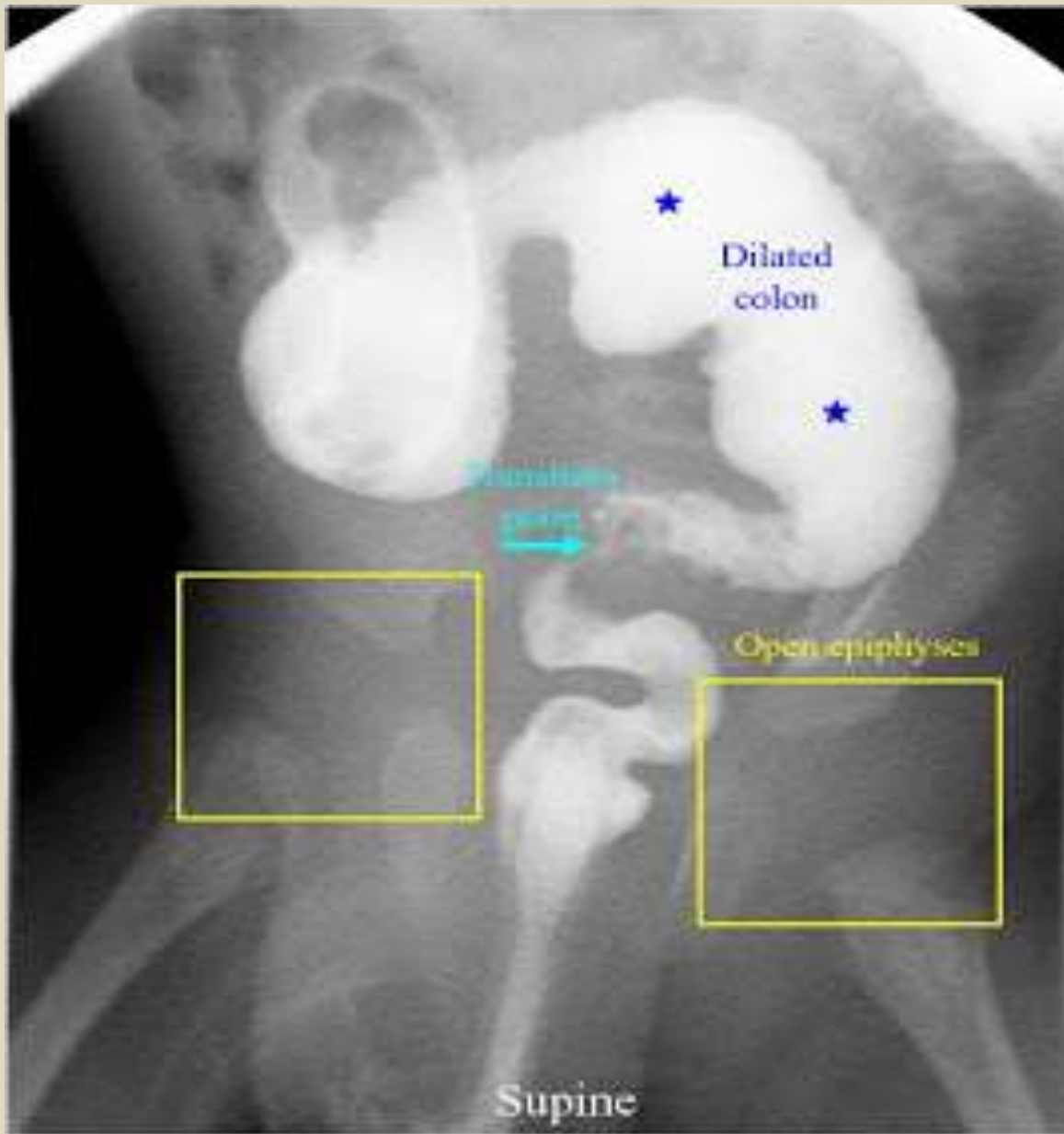
Hirschsprung disease. Barium enema showing reduced caliber of the rectum, followed by a transition zone to an enlarged-caliber sigmoid.



The transition zone is in the mid-descending colon.



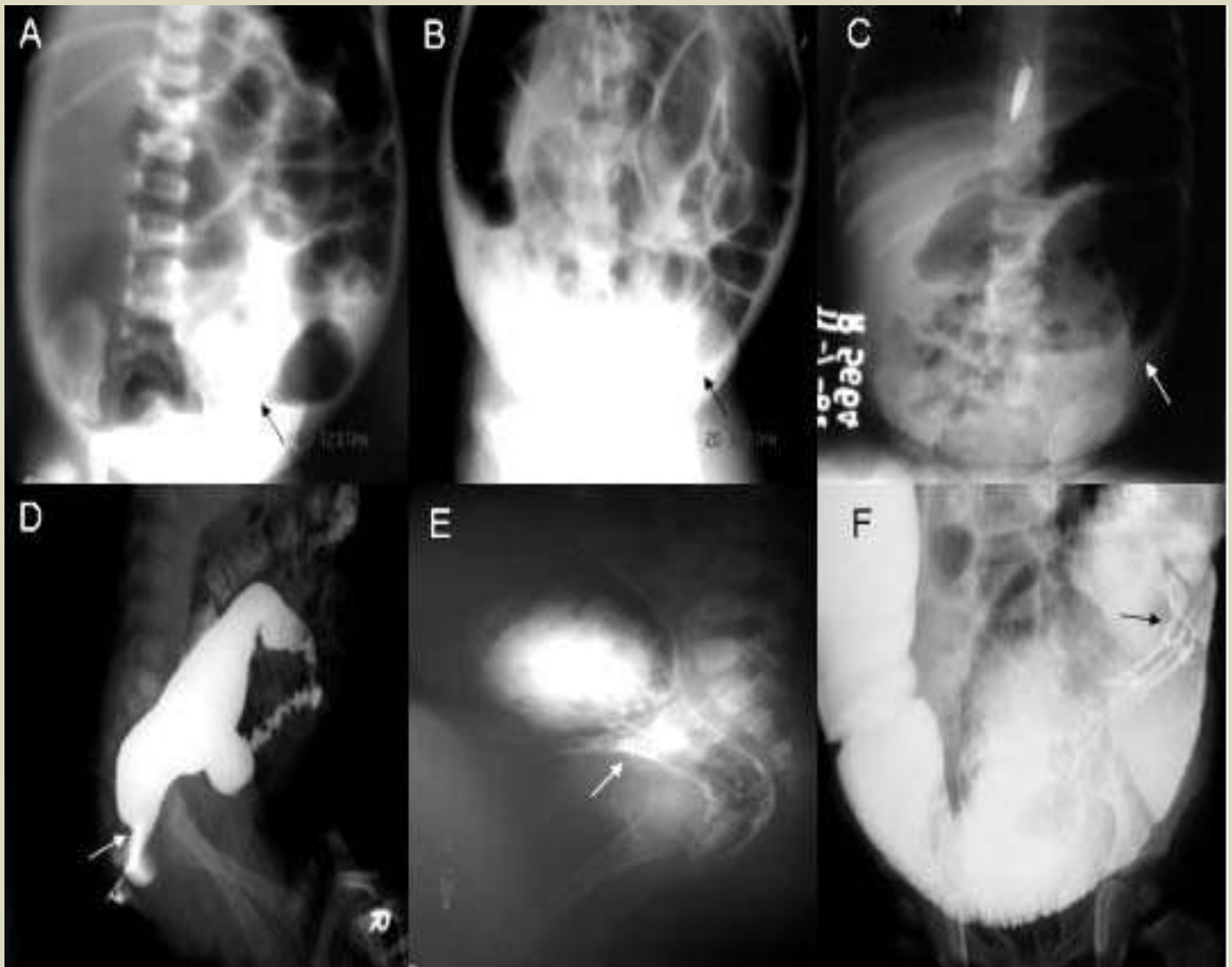
Hirschsprung disease. Barium enema showing reduced caliber and length of the large bowel, with no clear transition zone (total colonic aganglionosis).



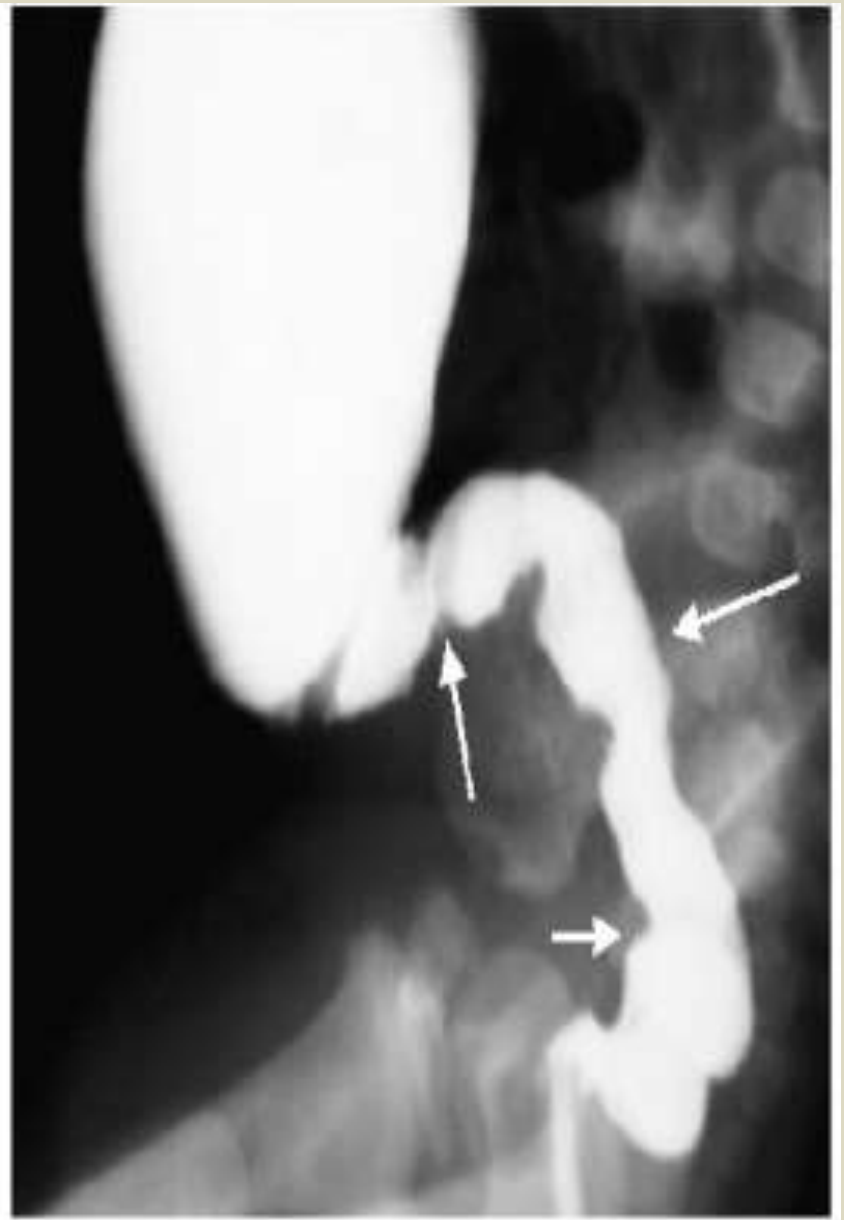
Hirschsprung Disease.



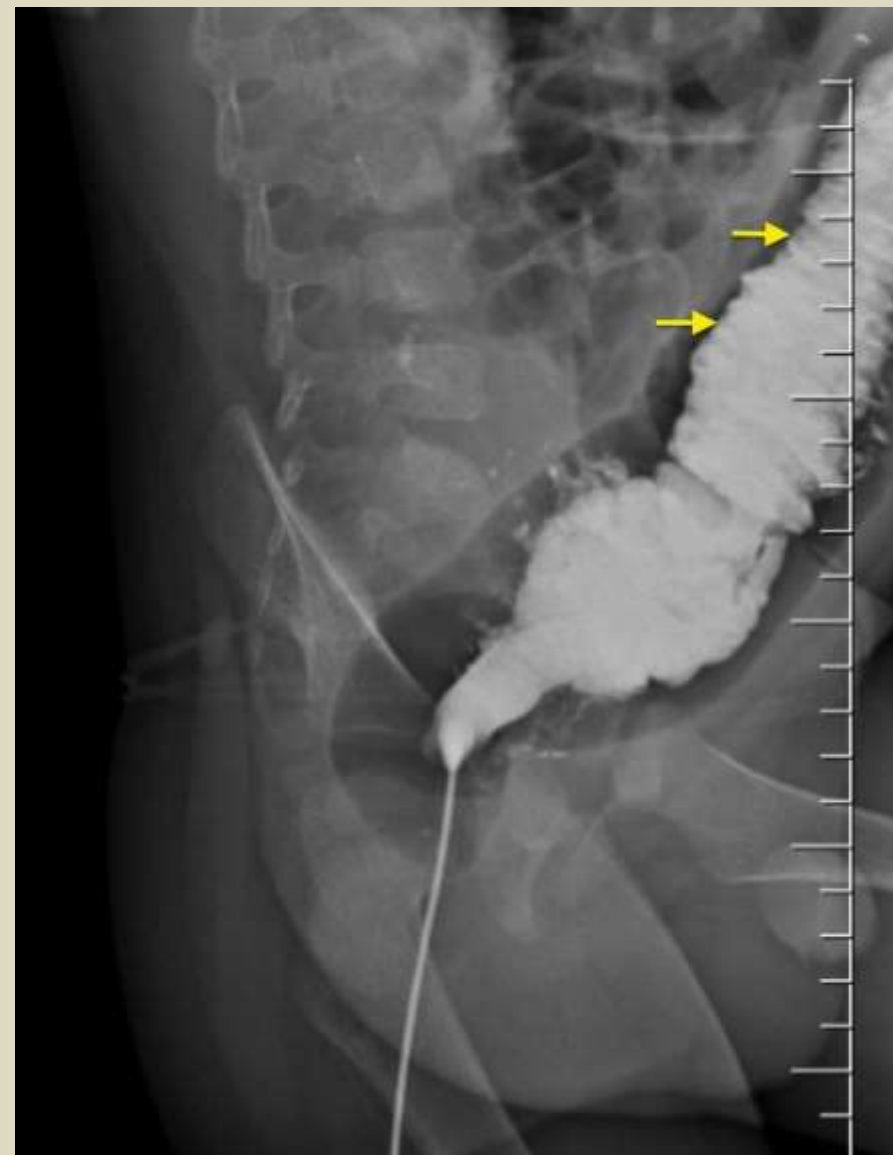
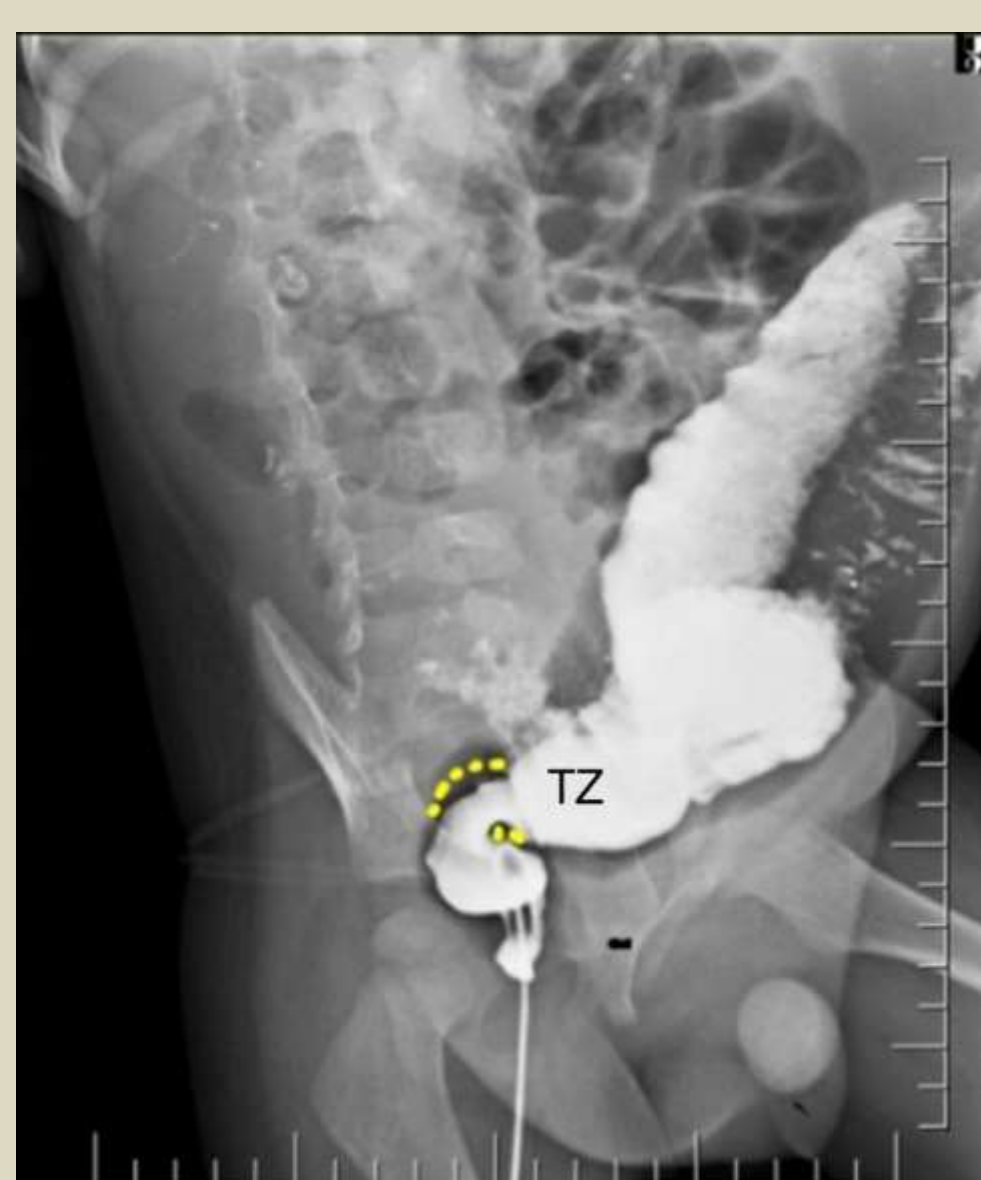
Hirschsprung disease in an infant. Frontal radiograph demonstrates the diameter of the rectum (arrows) to be smaller than the diameter of the sigmoid colon, an abnormal rectosigmoid ratio. Note the saw tooth appearance of the abnormal contracted segment.



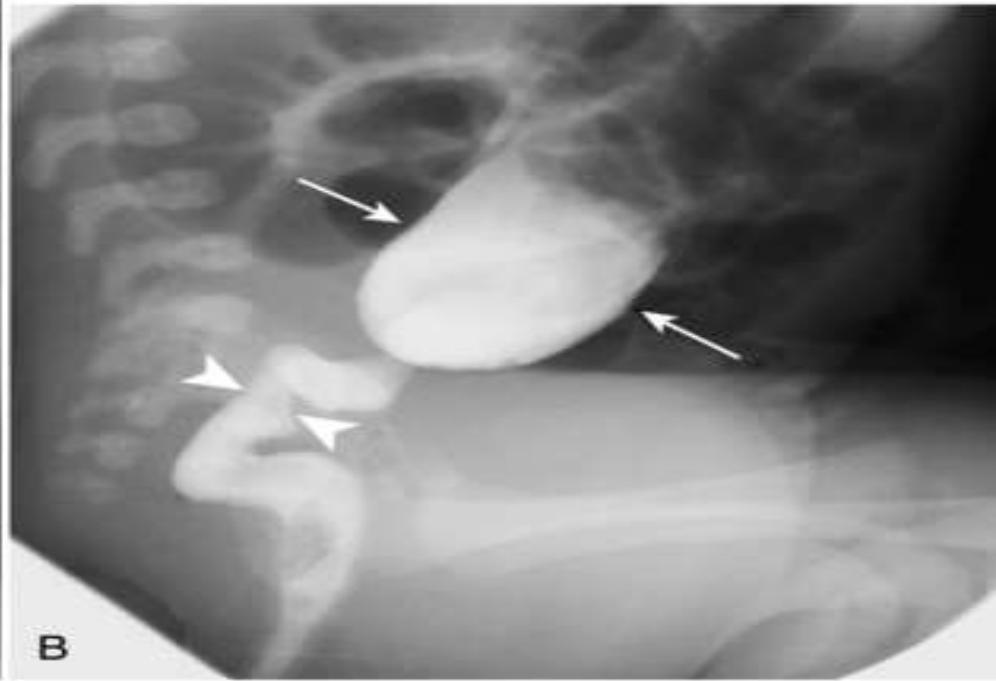
Hirschsprung Films A, B and C are plain abdominal x-rays. Films D, E and F are x-rays with a barium enema.



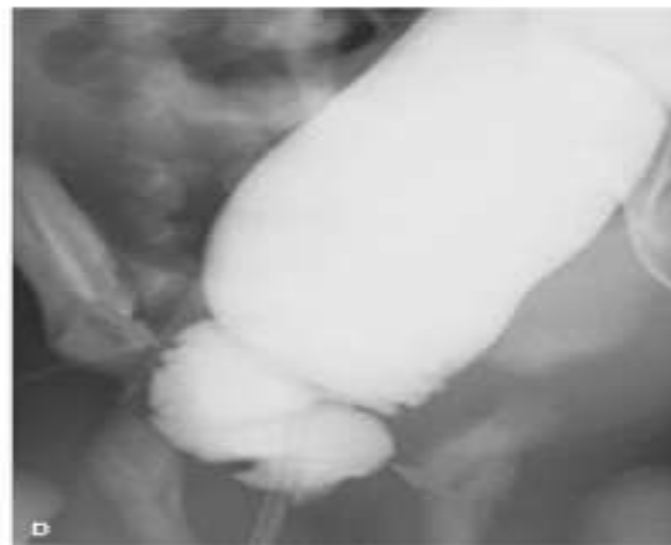
Hirschsprung Disease.



***Short narrowed segment indicated between the yellow dotted lines; TZ = transition zone
yellow arrows indicate the small bowel (jejunal) pattern to the descending colon***



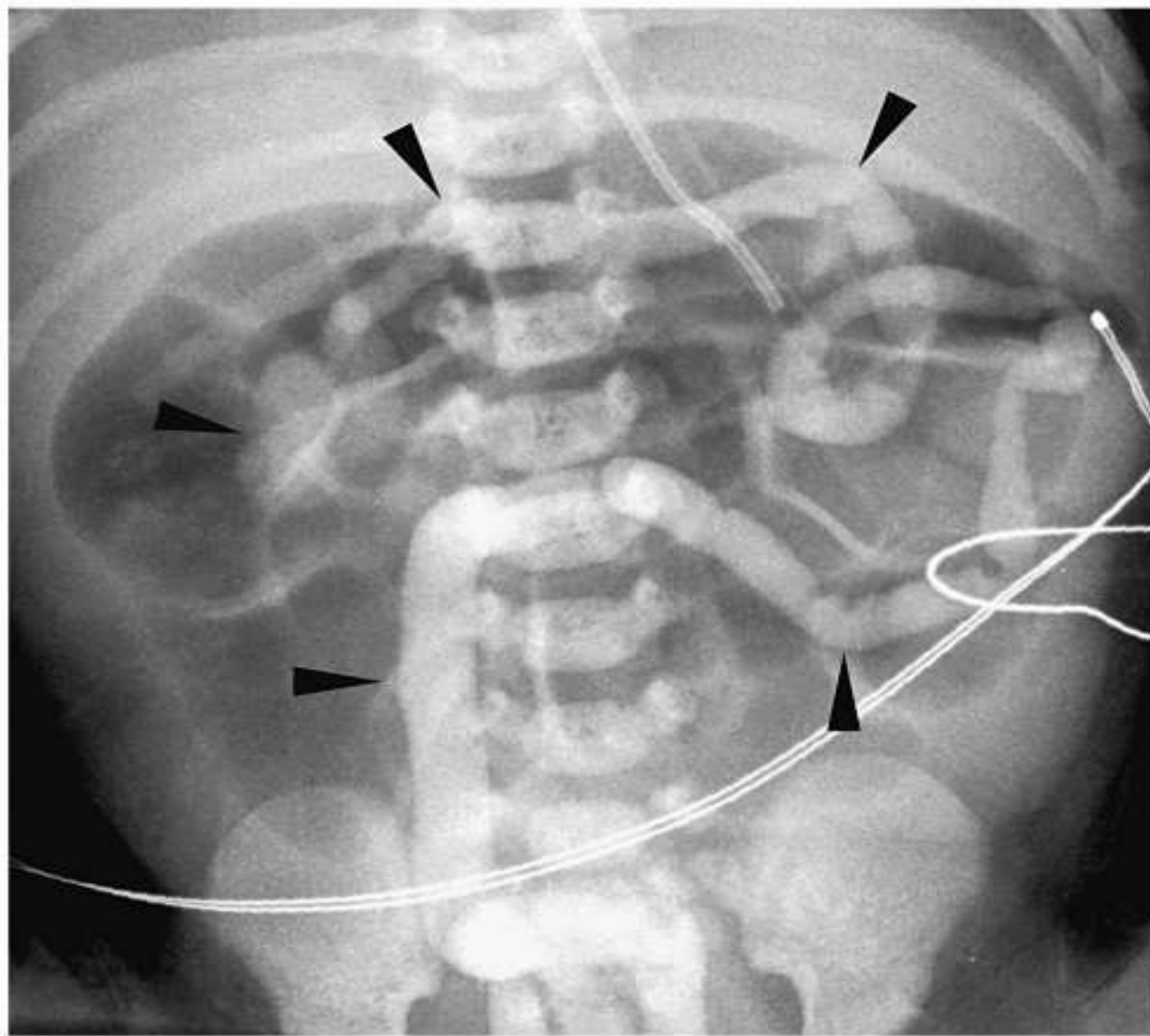
Hirschsprung disease in a newborn with abdominal distention and failure to pass meconium. A, Radiograph shows dilatation of multiple loops of bowel, consistent with a distal obstruction. B, Early filling lateral view from contrast enema shows abnormal rectosigmoid ratio with rectum (arrowheads) much more narrow than sigmoid colon (arrows). C, Frontal view from contrast enema shows rectum to be narrow in caliber and corkscrew in appearance due to spasm. The sigmoid colon (S) is dilated compared to the rectum (arrows).



Hirschsprung Disease. A. A characteristic transition zone (arrows) is seen between the dilated, feces-filled colon above and the relatively narrowed rectum below. B. The rectum in this newborn infant is smaller than the sigmoid and descending colon, but a well-defined transition zone is not present. C. A contrast enema in another infant shows spasm and irregularity of the aganglionic segment (arrow). D. A different infant with a shorter aganglionic segment (arrows) showing a mild corrugated pattern and a well-defined transition zone.

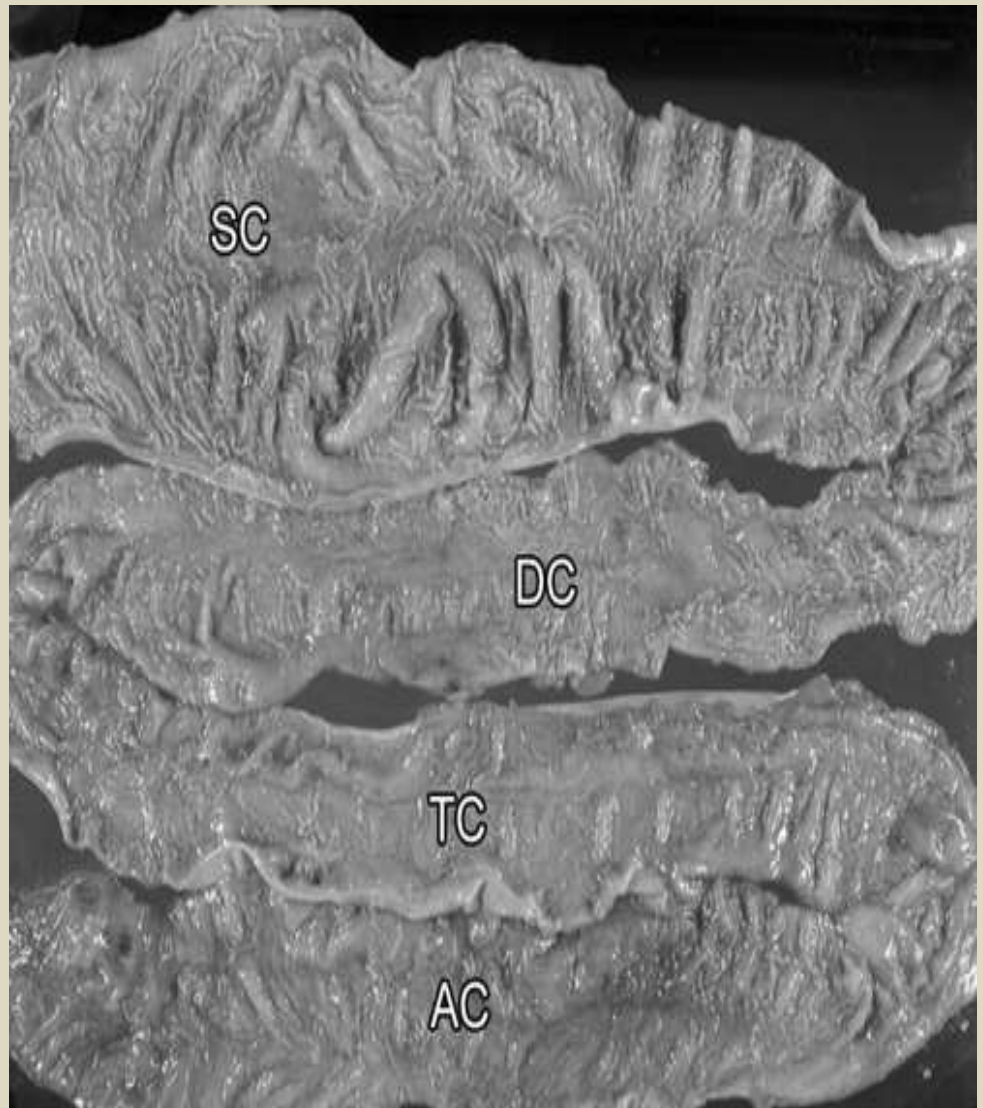
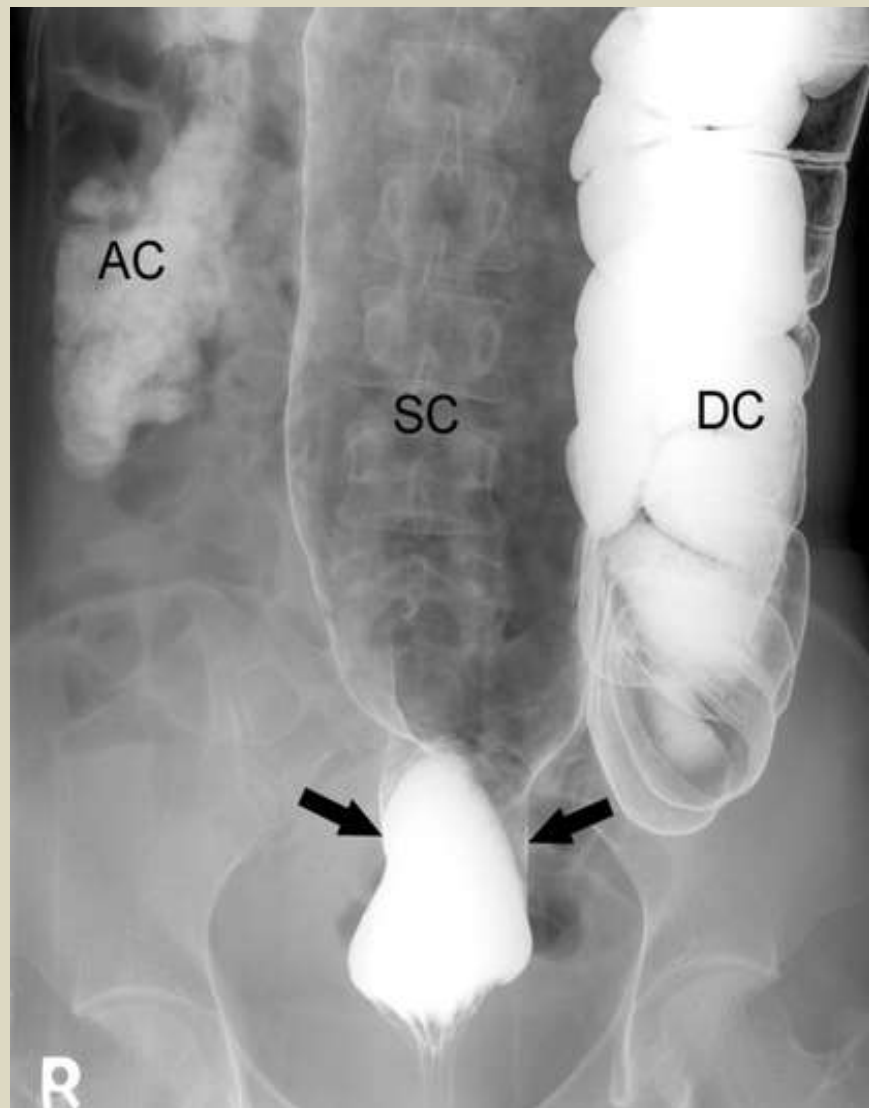


Total Colonic Aganglionosis—Long Segment Hirschsprung Disease .

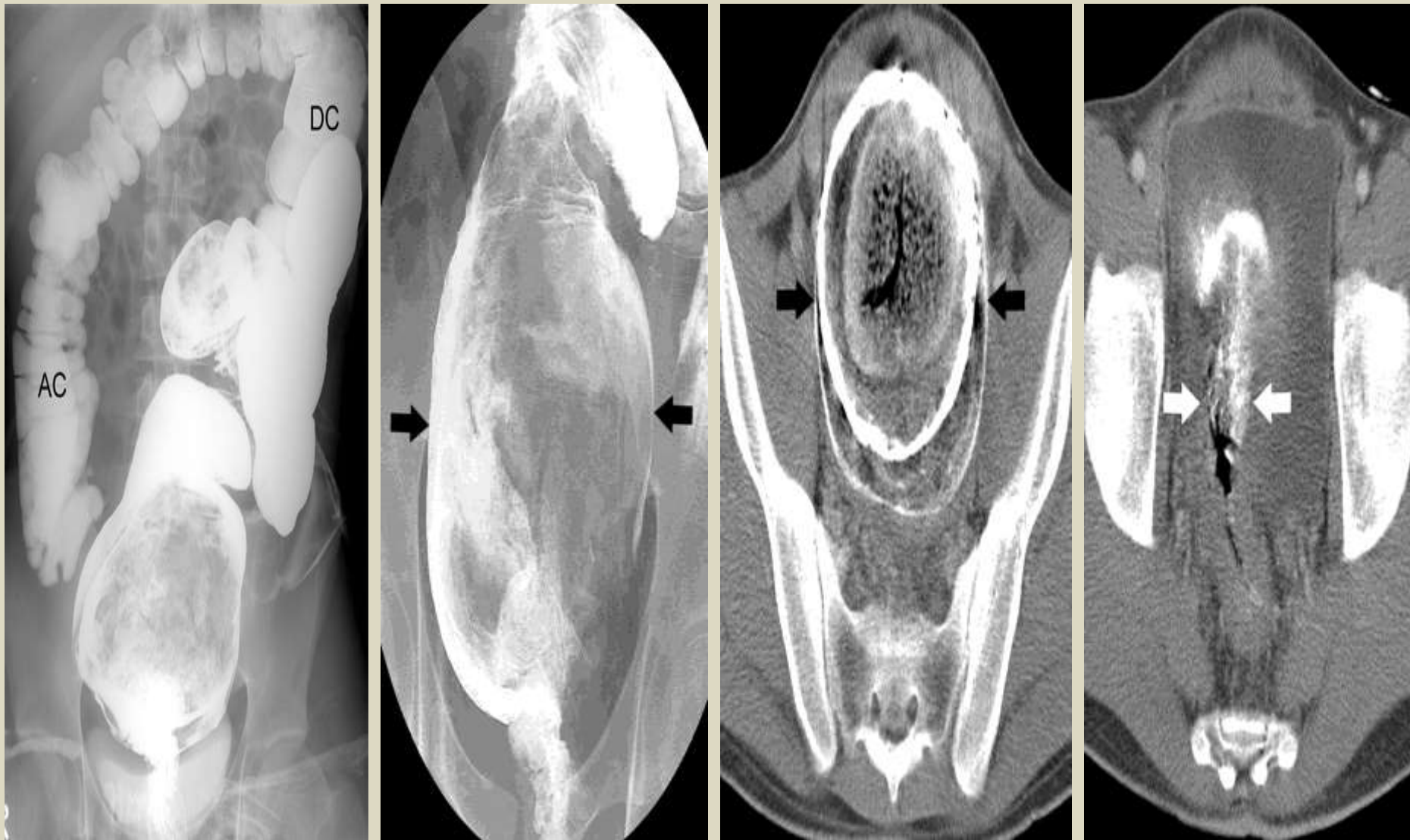


Total colonic Hirschsprung disease. The entire colon is small in caliber (arrowheads). The histologic transition zone was in the distal ileum.

Although CT is considered a common imaging modality for excluding other diseases such as colorectal cancer, which also causes chronic constipation in adults, to our knowledge, there have been no reports regarding the CT findings of adult HD and adult HG in the radiology literature. CT depicted the correct transition zone in nine patients with HG or HD, and the depicted transition zones were consistent with the histopathologic results. The transition zone was located in the rectum or rectosigmoid junction in all of the patients with HD. In addition, the transition zone in the patients with HG corresponded to the histopathologic results when it was considered to be in the segment with the least number of ganglion cells, as HG was distributed throughout the entire colon in four patients. These findings show that HG exhibits compensatory contraction like HD, even though, to our knowledge, this fact has not been mentioned in previous reports. There was a discrepancy between the transition zone site seen at histopathologic analysis and that seen at CT in one patient. The transition zone was in the distal transverse colon at histopathologic analysis but in the proximal descending colon at CT. This discrepancy was probably due to the redundancy of transverse and/or descending colon. A redundant colon depicted at transverse CT can make accurate localization of the transition zone difficult. Apparently, there seemed to be a short distance between the two transition zone sites. Also, the histopathologic type of combined HD and HG might have played a role in the discrepancy in transition zone sites.



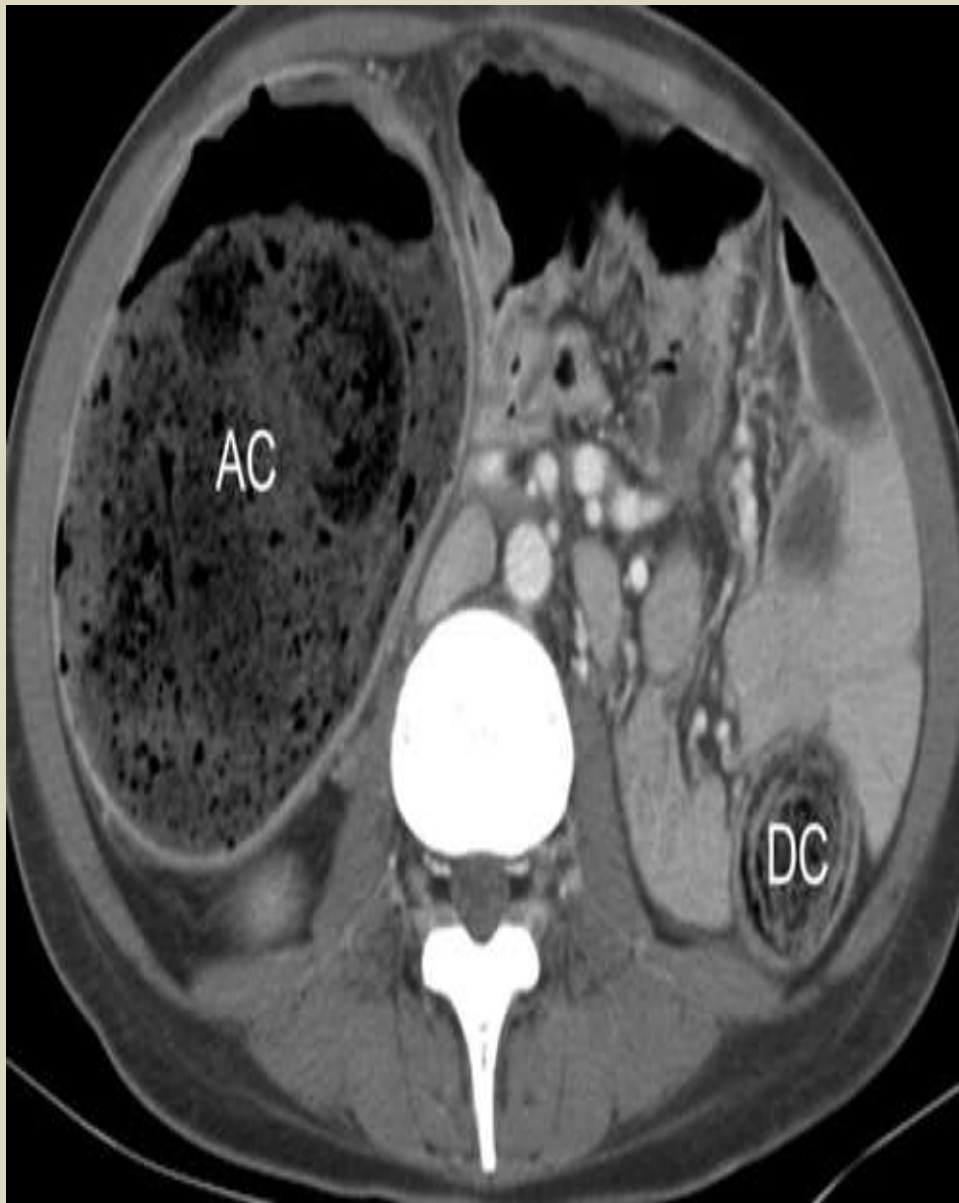
(a) Anteroposterior double-contrast barium enema radiograph (overhead view) shows mildly narrowed rectum and rectosigmoid junction (arrows) with dilated sigmoid colon (SC). AC = ascending colon, DC = descending colon. (b) Resected whole-colon specimen shows dilated sigmoid colon and non dilated ascending colon, transverse colon (TC), and descending colon.



HD with aganglionic segment of upper part of rectum in 19-year-old man. (a, b) Double-contrast barium enema radiographs (overhead and anteroposterior spot views) show a markedly dilated upper part of rectum and rectosigmoid junction with fecaloma (arrows). AC = ascending colon, DC = descending colon. Other colonic segments have normal diameters. (c, d) Contrast material-enhanced transverse CT scans show markedly dilated feces-filled proximal upper part of rectum and the rectosigmoid junction (arrows in c) with a transition zone and a distal narrowed rectum (arrows in d). The distal rectum also appears to have a thickened wall. The transition zone ratio is measured as the axial diameter at the most dilated proximal colonic segment (arrows in c) divided by the axial diameter at the narrowest distal part of the rectum (arrows in d).



Double-contrast barium enema radiographs (a, anteroposterior; b, oblique anteroposterior views) obtained in 64-year-old woman who has HD with aganglionic segment of rectosigmoid junction show markedly dilated distal sigmoid colon and rectosigmoid junction (arrowheads) and a narrowed rectum (arrows).



3a: Contrast-enhanced transverse CT scans (a obtained at lower level than b) obtained in 31-year-old woman with HG of entire colon show markedly dilated feces-filled ascending colon (AC) and transverse colon (arrowheads) compared with a nondilated descending colon (DC) with a transition zone of the proximal descending colon (*).

Thank You.